

Alessio Di Fonzo

List of Publications by Year in descending order

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Version: 2024-02-01

116
papers

8,140
citations

94269

37
h-index

51492

86
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121
all docs

121
docs citations

121
times ranked

10049
citing authors

#	ARTICLE	IF	CITATIONS
1	Guidelines for the use and interpretation of assays for monitoring autophagy (4th) Tj ETQq1 1 0.784314 rgBT /Overlock 10 Tf 50,742 1,430	4.3	1,430
2	Phenotype, genotype, and worldwide genetic penetrance of LRRK2-associated Parkinson's disease: a case-control study. <i>Lancet Neurology</i> , The, 2008, 7, 583-590.	4.9	1,340
3	A frequent LRRK2 gene mutation associated with autosomal dominant Parkinson's disease. <i>Lancet</i> , The, 2005, 365, 412-415.	6.3	449
4	<i>FBXO7</i> mutations cause autosomal recessive, early-onset parkinsonian-pyramidal syndrome. <i>Neurology</i> , 2009, 72, 240-245.	1.5	314
5	ATP13A2 missense mutations in juvenile parkinsonism and young onset Parkinson disease. <i>Neurology</i> , 2007, 68, 1557-1562.	1.5	312
6	Mutation in the<i>SYNJ1</i>Gene Associated with Autosomal Recessive, Early-Onset Parkinsonism. <i>Human Mutation</i> , 2013, 34, 1208-1215.	1.1	276
7	A common missense variant in the LRRK2 gene, Gly2385Arg, associated with Parkinsonâ€™s disease risk in Taiwan. <i>Neurogenetics</i> , 2006, 7, 133-138.	0.7	255
8	Adaptive deep brain stimulation in a freely moving parkinsonian patient. <i>Movement Disorders</i> , 2015, 30, 1003-1005.	2.2	198
9	GBA, Gaucher Disease, and Parkinsonâ€™s Disease: From Genetic to Clinic to New Therapeutic Approaches. <i>Cells</i> , 2019, 8, 364.	1.8	187
10	The Role of Mitochondria in Neurodegenerative Diseases: the Lesson from Alzheimerâ€™s Disease and Parkinsonâ€™s Disease. <i>Molecular Neurobiology</i> , 2020, 57, 2959-2980.	1.9	180
11	The G6055A (G2019S) mutation in LRRK2 is frequent in both early and late onset Parkinson's disease and originates from a common ancestor. <i>Journal of Medical Genetics</i> , 2005, 42, e65-e65.	1.5	178
12	The LRRK2 Gly2385Arg variant is associated with Parkinsonâ€™s disease: genetic and functional evidence. <i>Human Genetics</i> , 2007, 120, 857-863.	1.8	157
13	Comprehensive analysis of the LRRK2 gene in sixty families with Parkinson's disease. <i>European Journal of Human Genetics</i> , 2006, 14, 322-331.	1.4	152
14	The Mitochondrial Disulfide Relay System Protein GFER Is Mutated in Autosomal-Recessive Myopathy with Cataract and Combined Respiratory-Chain Deficiency. <i>American Journal of Human Genetics</i> , 2009, 84, 594-604.	2.6	121
15	Mutations in DNA2 Link Progressive Myopathy to Mitochondrial DNA Instability. <i>American Journal of Human Genetics</i> , 2013, 92, 293-300.	2.6	115
16	Cerebellar tDCS: How to Do It. <i>Cerebellum</i> , 2015, 14, 27-30.	1.4	114
17	LRRK2 G2019S mutation and Parkinson's disease: A clinical, neuropsychological and neuropsychiatric study in a large Italian sample. <i>Parkinsonism and Related Disorders</i> , 2006, 12, 410-419.	1.1	106
18	LRP10 genetic variants in familial Parkinson's disease and dementia with Lewy bodies: a genome-wide linkage and sequencing study. <i>Lancet Neurology</i> , The, 2018, 17, 597-608.	4.9	101

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19	POLG mutations in sporadic mitochondrial disorders with multiple mtDNA deletions. <i>Human Mutation</i> , 2003, 22, 498-499.	1.1	100
20	Cerebellar and Motor Cortical Transcranial Stimulation Decrease Levodopa-Induced Dyskinesias in Parkinson's Disease. <i>Cerebellum</i> , 2016, 15, 43-47.	1.4	99
21	Adaptive deep brain stimulation controls levodopa-induced side effects in Parkinsonian patients. <i>Movement Disorders</i> , 2017, 32, 628-629.	2.2	96
22	Novel ATP13A2 (PARK9) homozygous mutation in a family with marked phenotype variability. <i>Neurogenetics</i> , 2011, 12, 33-39.	0.7	84
23	GBA-Related Parkinson's Disease: Dissection of Genotype-Phenotype Correlates in a Large Italian Cohort. <i>Movement Disorders</i> , 2020, 35, 2106-2111.	2.2	83
24	High prevalence of LRRK2 mutations in familial and sporadic Parkinson's disease in Portugal. <i>Movement Disorders</i> , 2007, 22, 1194-1201.	2.2	76
25	The LRRK2 I2012T, G2019S, and I2020T mutations are rare in Taiwanese patients with sporadic Parkinson's disease. <i>Parkinsonism and Related Disorders</i> , 2005, 11, 521-522.	1.1	70
26	Leucine-Rich Repeat Kinase (LRRK2) Genetics and Parkinson's Disease. <i>Advances in Neurobiology</i> , 2017, 14, 3-30.	1.3	66
27	The GBAP1 pseudogene acts as a ceRNA for the glucocerebrosidase gene GBA by sponging miR-22-3p. <i>Scientific Reports</i> , 2017, 7, 12702.	1.6	62
28	The LRRK2 Arg1628Pro variant is a risk factor for Parkinson's disease in the Chinese population. <i>Neurogenetics</i> , 2008, 9, 271-276.	0.7	61
29	Remarkable infidelity of polymerase β associated with mutations in POLG1 exonuclease domain. <i>Neurology</i> , 2003, 61, 903-908.	1.5	60
30	Novel mitochondrial protein interactors of immunoglobulin light chains causing heart amyloidosis. <i>FASEB Journal</i> , 2015, 29, 4614-4628.	0.2	60
31	Understanding the pathogenesis of multiple system atrophy: state of the art and future perspectives. <i>Acta Neuropathologica Communications</i> , 2019, 7, 113.	2.4	56
32	Neuropathology of Parkinson's disease associated with the LRRK2 Ile1371Val mutation. <i>Movement Disorders</i> , 2007, 22, 275-278.	2.2	46
33	Mitochondrial Dysregulation and Impaired Autophagy in iPSC-Derived Dopaminergic Neurons of Multiple System Atrophy. <i>Stem Cell Reports</i> , 2018, 11, 1185-1198.	2.3	46
34	Autophagy in motor neuron disease: Key pathogenetic mechanisms and therapeutic targets. <i>Molecular and Cellular Neurosciences</i> , 2016, 72, 84-90.	1.0	43
35	X-linked Parkinsonism with Intellectual Disability caused by novel mutations and somatic mosaicism in RAB39B gene. <i>Parkinsonism and Related Disorders</i> , 2017, 44, 142-146.	1.1	43
36	Striatal dopamine transporter binding in Parkinson's disease associated with the LRRK2 Gly2019Ser mutation. <i>Movement Disorders</i> , 2006, 21, 1144-1147.	2.2	41

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37	SPG11: a consistent clinical phenotype in a family with homozygous Spatacsin truncating mutation. <i>Neurogenetics</i> , 2007, 8, 301-305.	0.7	38
38	Motor and cognitive outcomes of cerebello-spinal stimulation in neurodegenerative ataxia. <i>Brain</i> , 2021, 144, 2310-2321.	3.7	38
39	The SPID-GBA study. <i>Neurology: Genetics</i> , 2020, 6, e523.	0.9	37
40	Nucleoâ€cytoplasmic transport defects and protein aggregates in neurodegeneration. <i>Translational Neurodegeneration</i> , 2020, 9, 25.	3.6	33
41	Novel missense mutation and large deletion of GNE gene in autosomal-recessive inclusion-body myopathy. <i>Muscle and Nerve</i> , 2003, 28, 113-117.	1.0	32
42	Mitochondrial dysfunction in fibroblasts of Multiple System Atrophy. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2018, 1864, 3588-3597.	1.8	32
43	Spinal direct current stimulation (tsDCS) in hereditary spastic paraplegias (HSP): A sham-controlled crossover study. <i>Journal of Spinal Cord Medicine</i> , 2021, 44, 46-53.	0.7	29
44	Discrimination of MSA-P and MSA-C by RT-QuIC analysis of olfactory mucosa: the first assessment of assay reproducibility between two specialized laboratories. <i>Molecular Neurodegeneration</i> , 2021, 16, 82.	4.4	28
45	Genetics of Movement Disorders and the Practicing Clinician; Who and What to Test for?. <i>Current Neurology and Neuroscience Reports</i> , 2018, 18, 37.	2.0	27
46	Mutational analysis of COQ2 in patients with MSA in Italy. <i>Neurobiology of Aging</i> , 2016, 45, 213.e1-213.e2.	1.5	25
47	Neurofascin (NFASC) gene mutation causes autosomal recessive ataxia with demyelinating neuropathy. <i>Parkinsonism and Related Disorders</i> , 2019, 63, 66-72.	1.1	25
48	Obesity and Headache/Migraine: The Importance of Weight Reduction through Lifestyle Modifications. <i>BioMed Research International</i> , 2014, 2014, 1-7.	0.9	24
49	GIGYF2 mutations are not a frequent cause of familial Parkinson's disease. <i>Parkinsonism and Related Disorders</i> , 2009, 15, 703-705.	1.1	22
50	A novel homozygous PLA2G6 mutation causes dystonia-parkinsonism. <i>Parkinsonism and Related Disorders</i> , 2015, 21, 337-339.	1.1	22
51	Real life evaluation of safinamide effectiveness in Parkinsonâ€™s disease. <i>Neurological Sciences</i> , 2018, 39, 733-739.	0.9	22
52	LRRK2 mutations and Parkinson's disease in Sardiniaâ€™A Mediterranean genetic isolate. <i>Parkinsonism and Related Disorders</i> , 2007, 13, 17-21.	1.1	21
53	Lower motor neuron disease with respiratory failure caused by a novel <i>MAPT</i> mutation. <i>Neurology</i> , 2014, 82, 1990-1998.	1.5	21
54	The Length of SNCA Rep1 Microsatellite May Influence Cognitive Evolution in Parkinsonâ€™s Disease. <i>Frontiers in Neurology</i> , 2018, 9, 213.	1.1	21

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55	HOPS-associated neurological disorders (HOPSANDs): linking endolysosomal dysfunction to the pathogenesis of dystonia. <i>Brain</i> , 2021, 144, 2610-2615.	3.7	20
56	Unusual adult-onset Leigh syndrome presentation due to the mitochondrial m.9176T>C mutation. <i>Biochemical and Biophysical Research Communications</i> , 2011, 412, 245-248.	1.0	19
57	Targeting the Autonomic Nervous System for Risk Stratification, Outcome Prediction and Neuromodulation in Ischemic Stroke. <i>International Journal of Molecular Sciences</i> , 2021, 22, 2357.	1.8	19
58	A Practical Approach to Early-Onset Parkinsonism. <i>Journal of Parkinson's Disease</i> , 2022, 12, 1-26.	1.5	19
59	Progressive Encephalomyelitis with Rigidity and Myoclonus Associated With Anti-GlyR Antibodies and Hodgkin's Lymphoma: A Case Report. <i>Frontiers in Neurology</i> , 2017, 8, 401.	1.1	17
60	Role of Lysosomal Gene Variants in Modulating <i>GBA</i> -Associated Parkinson's Disease Risk. <i>Movement Disorders</i> , 2022, 37, 1202-1210.	2.2	17
61	SLC25A46 mutations in patients with Parkinson's Disease and optic atrophy. <i>Parkinsonism and Related Disorders</i> , 2020, 74, 1-5.	1.1	16
62	A case of CPT deficiency, homoplasmic mtDNA mutation and ragged red fibers at muscle biopsy. <i>Journal of the Neurological Sciences</i> , 2005, 239, 21-24.	0.3	15
63	Parkin polymorphisms and environmental exposure: Decrease in age at onset of Parkinson's disease. <i>NeuroToxicology</i> , 2007, 28, 698-701.	1.4	15
64	A de novo C19orf12 heterozygous mutation in a patient with MPAN. <i>Parkinsonism and Related Disorders</i> , 2018, 48, 109-111.	1.1	15
65	Leukoencephalopathy with calcifications and cysts: Genetic and phenotypic spectrum. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 15-25.	0.7	15
66	Abnormal brain temperature in early-onset Parkinson's disease. <i>Movement Disorders</i> , 2016, 31, 425-426.	2.2	14
67	Congenital Myasthenic Syndrome Due to Choline Acetyltransferase Mutations in Infants. <i>Journal of Child Neurology</i> , 2014, 29, 389-393.	0.7	13
68	Pharmacological antagonism of kainate receptor rescues dysfunction and loss of dopamine neurons in a mouse model of human parkin-induced toxicity. <i>Cell Death and Disease</i> , 2020, 11, 963.	2.7	13
69	A Novel Homozygous <i>VPS11</i> Variant May Cause Generalized Dystonia. <i>Annals of Neurology</i> , 2021, 89, 834-839.	2.8	13
70	Genetic variants in levodopa-induced dyskinesia (LID): A systematic review and meta-analysis. <i>Parkinsonism and Related Disorders</i> , 2021, 84, 52-60.	1.1	13
71	Autosomal dominant restless legs syndrome maps to chromosome 20p13 (<i>RLS5</i>) in a Dutch kindred. <i>Movement Disorders</i> , 2010, 25, 1715-1722.	2.2	12
72	The Italian tremor Network (TITAN): rationale, design and preliminary findings. <i>Neurological Sciences</i> , 2022, 43, 5369-5376.	0.9	12

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73	Mutations in TMEM230 are rare in autosomal dominant Parkinson's disease. <i>Parkinsonism and Related Disorders</i> , 2017, 39, 87-88.	1.1	11
74	In vitro models of multiple system atrophy from primary cells to induced pluripotent stem cells. <i>Journal of Cellular and Molecular Medicine</i> , 2018, 22, 2536-2546.	1.6	11
75	<i>LRRK2</i> MUTATION ANALYSIS IN PARKINSON DISEASE FAMILIES WITH EVIDENCE OF LINKAGE TO PARK8. <i>Neurology</i> , 2008, 70, 2348-2349.	1.5	10
76	Pseudoorthostatic and resting leg tremor in a large spanish family with homozygous truncating <i>parkin</i> mutation. <i>Movement Disorders</i> , 2009, 24, 144-147.	2.2	10
77	Juvenile dystonia-parkinsonism syndrome caused by a novel p.S941Tfs1X ATP13A2 (PARK9) mutation. <i>Parkinsonism and Related Disorders</i> , 2015, 21, 1378-1380.	1.1	10
78	Dystonia-ataxia syndrome with permanent torsional nystagmus caused by ECHS1 deficiency. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 839-845.	1.7	10
79	<i>TWINK</i> in Parkinson's Disease: A Movement Disorder and Mitochondrial Disease Center Perspective Study. <i>Movement Disorders</i> , 2022, 37, 1938-1943.	2.2	10
80	Globus pallidus internus deep brain stimulation in PINK-1 related Parkinson's disease: A case report. <i>Parkinsonism and Related Disorders</i> , 2017, 38, 93-94.	1.1	9
81	Loss of the nucleoporin Aladin in central nervous system and fibroblasts of Allgrove Syndrome. <i>Human Molecular Genetics</i> , 2019, 28, 3921-3927.	1.4	9
82	Unravelling Genetic Factors Underlying Corticobasal Syndrome: A Systematic Review. <i>Cells</i> , 2021, 10, 171.	1.8	8
83	VPS13C-associated Parkinson's disease: Two novel cases and review of the literature. <i>Parkinsonism and Related Disorders</i> , 2022, 94, 37-39.	1.1	8
84	Two novel mutations in PEO1 (Twinkle) gene associated with chronic external ophthalmoplegia. <i>Journal of the Neurological Sciences</i> , 2011, 308, 173-176.	0.3	7
85	A rapid and low-cost test for screening the most common Parkinson's disease-related GBA variants. <i>Parkinsonism and Related Disorders</i> , 2020, 80, 138-141.	1.1	7
86	Systemic involvement in adult-onset leukoencephalopathy with intracranial calcifications and cysts (Labrune syndrome) with a novel mutation of the SNORD118 gene. <i>European Journal of Neurology</i> , 2020, 27, 2329-2332.	1.7	6
87	Dysautonomia in Parkinson's Disease: Impact of Glucocerebrosidase Gene Mutations on Cardiovascular Autonomic Control. <i>Frontiers in Neuroscience</i> , 2022, 16, 842498.	1.4	6
88	Validation of the Italian version of the PSP Quality of Life questionnaire. <i>Neurological Sciences</i> , 2019, 40, 2587-2594.	0.9	5
89	Validation of the Italian version of carers' quality-of-life questionnaire for parkinsonism (PQoL). <i>TJ ETQq1</i> 1 0.784314 rgBT ₅ / Overlook	0.9	5
90	Screening of LRP10 mutations in Parkinson's disease patients from Italy. <i>Parkinsonism and Related Disorders</i> , 2021, 89, 17-21.	1.1	5

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91	The novel mitochondrial tRNA ^{Asn} gene mutation m.5709T>C produces ophthalmoparesis and respiratory impairment. <i>European Journal of Human Genetics</i> , 2012, 20, 357-360.	1.4	4
92	Parkinsonism in diseases predominantly presenting with dystonia. <i>International Review of Neurobiology</i> , 2019, 149, 307-326.	0.9	4
93	Progressive myoclonus without epilepsy due to a NUS1 frameshift insertion: Dyssynergia cerebellaris myoclonica revisited. <i>Parkinsonism and Related Disorders</i> , 2022, 98, 53-55.	1.1	4
94	Clinical uses of Bupropion in patients with Parkinson's disease and comorbid depressive or neuropsychiatric symptoms: a scoping review. <i>BMC Neurology</i> , 2022, 22, 169.	0.8	4
95	Parkinson's disease in Gaucher disease patients: what's changing in the counseling and management of patients and their relatives?. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 262.	1.2	3
96	Comprehensive Genomic Analysis Reveals the Prognostic Role of LRRK2 Copy-Number Variations in Human Malignancies. <i>Genes</i> , 2020, 11, 846.	1.0	3
97	A case report of late-onset cerebellar ataxia associated with a rare p.R342W TGM6 (SCA35) mutation. <i>BMC Neurology</i> , 2020, 20, 408.	0.8	3
98	Design and Operation of the Lombardy Parkinson's Disease Network. <i>Frontiers in Neurology</i> , 2020, 11, 573.	1.1	3
99	Microscopic Polyangiitis With Selective Involvement of Central and Peripheral Nervous System: A Case Report. <i>Frontiers in Neurology</i> , 2020, 11, 269.	1.1	3
100	Clinical Reasoning: A 75-year-old man with parkinsonism, mood depression, and weight loss. <i>Neurology</i> , 2018, 90, 572-575.	1.5	2
101	Sodium Levels Predict Disability at Discharge in Guillain-Barré Syndrome: A Retrospective Cohort Study. <i>Frontiers in Neurology</i> , 2021, 12, 729252.	1.1	2
102	Parkinsonism and ataxia. <i>Journal of the Neurological Sciences</i> , 2021, , 120020.	0.3	2
103	Childhood-onset dystonia with cerebellar signs: expanding the spectrum of GNAL mutations. <i>European Journal of Neurology</i> , 2020, 27, e66-e67.	1.7	1
104	Transcriptomic characterization of tissues from patients and subsequent pathway analyses reveal biological pathways that are implicated in spastic ataxia. <i>Cell and Bioscience</i> , 2022, 12, 29.	2.1	1
105	1.283 Clinical and genetic study of a large Dutch family with autosomal dominant restless legs syndrome. <i>Parkinsonism and Related Disorders</i> , 2007, 13, S72.	1.1	0
106	2.119 Parkin polymorphisms and environmental exposure: Reduction of Parkinson's disease age of onset. <i>Parkinsonism and Related Disorders</i> , 2007, 13, S97.	1.1	0
107	Generation and characterization of iPSC-derived cortical pyramidal neurons from patients affected by multiple system atrophy. <i>Parkinsonism and Related Disorders</i> , 2016, 22, e119-e120.	1.1	0
108	LRRK2 and GBA mutation analysis in a cohort of Italian familial and sporadic PD. <i>Parkinsonism and Related Disorders</i> , 2016, 22, e167.	1.1	0

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109	Hallucinations in Neurological Disorders. , 2018, , 99-130.		0
110	Syncope and autonomic failure in a middle-aged man. Internal and Emergency Medicine, 2019, 14, 271-274.	1.0	0
111	Late-onset leukoencephalopathy in a patient with recessive EARS2 mutations. Neurology: Genetics, 2020, 6, e488.	0.9	0
112	Expanding the genotypic and phenotypic spectrum of Betaâ€propeller poteinâ€associated neurodegeneration. European Journal of Neurology, 2021, 28, e25-e27.	1.7	0
113	Clinical features and disease course of patients with acute ischaemic stroke just before the Italian index case: Was COVID-19 already there?. Internal and Emergency Medicine, 2021, 16, 1247-1252.	1.0	0
114	A 79-year-old man with unexplained recurrent syncope and severe orthostatic hypotension. Internal and Emergency Medicine, 2021, , 1.	1.0	0
115	The SPID- <i>GBA</i> Study: The Largest Monocentric Study on Sex Distribution, Penetrance, Incidence, and Association with Dementia of <i>GBA</i> Mutations in Parkinson's Disease. SSRN Electronic Journal, 0, , .	0.4	0
116	Genetic evaluation in phenotypically discordant monozygotic twins with Coats Disease. European Journal of Ophthalmology, 0, , 112067212211077.	0.7	0